

DISORDERS COVERED BY THE PROGRAM

Effective January 1, 2006, Utah newborns are screened for the following disorders:

METABOLIC DISORDERS:

- Biotinidase deficiency
- Galactosemia

Fatty Acid Oxidation Disorders:

- Carnitine uptake/transport defects
- Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Long chain 3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Carnitine-Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase-1 Deficiency

Amino acid disorders:

- Arginase Deficiency
- Argininosuccinate lyase deficiency (ASA)
- Citrullinemia
- Homocystinuria
- Hyperphenylalanemia, including phenylketonuria
- Tyrosinemia

Organic Acid Disorders:

- Beta-ketothiolase deficiency
- Glutaric acidemia, Type 1
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleric acidemia
- Malonic aciduria
- Maple syrup urine disease
- Methylmalonic acidemias
- Propionic acidemia
- 3-Hydroxy-3-methylglutaryl (HMG) CoA lyase deficiency
- 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency
- 2-Methylbutyryl CoA dehydrogenase deficiency
- Multiple carboxylase deficiency

ENDOCRINE DISORDERS:

- Congenital adrenal hyperplasia (CAH)
- Congenital hypothyroidism

HEMOGLOBIN DISORDERS:

- Sickle cell disease and other hemoglobinopathies

These are disorders which may have significant mortality and morbidity when not diagnosed presymptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders.



Newborn
Screening Program

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